共催：日本人類遺伝学会 第71回大会／株式会社ジーンベイ

スイーツセミナー

希少疾患診断を加速するナノポアシーケンスの可能性

2026 11.19 THU. 15:00–16:00

整理券、事前予約不要！

会　場　仙台国際センター展示棟 第5会場（会議室2）

共　催　株式会社ジーンベイ

座　長　上村 泰央（株式会社ジーンベイ）

Analysis of complex chromosomal structural abnormalities using nanopore long-read sequencing

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Chromosomal microarray testing (CMT) is a powerful tool to detect copy number variations (CNVs); however, we sometimes experience complex findings that cannot be explained by simple deletions or duplications. Since CMT can only detect the existence of CNVs, actual genomic structures of them cannot be detected. Cytogenetic techniques—specifically FISH—can be used to verify structural details in cases involving, for instance, subtelomeric translocations. However, confirmation via FISH is difficult when the regions involved are not amenable to commercial FISH testing or when the CNVs affect very small genomic segments. In such cases, nanopore long-read DNA sequencing has advantages. CMT results initially interpreted as three independent deletions or as a duplication superimposed on another duplication were actually complex structural abnormalities resulting from processes such as chromothripsis or chromoanasynthesis. Conversely, nanopore long-read DNA sequencing also can be used to detect cryptic structural changes in the non-coding regions. In cases where possible candidate genes were narrowed down based on characteristic clinical symptoms but no single-nucleotide variants (SNVs) were found through whole exome sequencing, nanopore long-read DNA sequencing may detect genomic alterations in the deep intronic regions. Here, we present cases in which nanopore long-read DNA sequencing successfully identified the structural abnormalities responsible for the disease.

会期中は企業展示も行っております。ぜひお越しください！

日　時　11月19日（木）9:00－17:30
11月20日（金）9:00－17:00
11月21日（土）9:00－16:00

会　場　仙台国際センター展示棟
〈ブース番号〉19

出展内容
・ナノポアシークエンスおよび各種NGS受託解析
・GPU解析システム（ナノポアベースコール解析および各種解析パイプライン）



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